

RAW SEQUENCE LISTING

**The Biotechnology Systems Branch of the Scientific and Technical
Information Center (STIC) no errors detected.**

Application Serial Number: 10/560,157
Source: IFWP
Date Processed by STIC: 12/20/2005

ENTERED



IFWP

RAW SEQUENCE LISTING

DATE: 12/20/2005

PATENT APPLICATION: US/10/560,157

TIME: 11:17:28

Input Set : A:\Seqlist.txt

Output Set: N:\CRF4\12202005\J560157.raw

3 <110> APPLICANT: Pietrangelo, Antonello
 5 <120> TITLE OF INVENTION: Mutations in the SLC40A1 gene associated to
 impaired iron homeostasis
 7 <130> FILE REFERENCE: 8907-109-999
C--> 9 <140> CURRENT APPLICATION NUMBER: US/10/560,157
 10 <141> CURRENT FILING DATE: 2005-12-09
 12 <150> PRIOR APPLICATION NUMBER: PCT/EP2004/051068
 13 <151> PRIOR FILING DATE: 2004-06-09
 15 <160> NUMBER OF SEQ ID NOS: 30
 17 <170> SOFTWARE: PatentIn version 3.1
 19 <210> SEQ ID NO: 1
 20 <211> LENGTH: 1716
 21 <212> TYPE: DNA
 22 <213> ORGANISM: Homo sapiens
 24 <220> FEATURE:
 25 <221> NAME/KEY: CDS
 26 <222> LOCATION: (1)..(1716)
 27 <223> OTHER INFORMATION: cDNA encoding wild type ferroportin 1.
 Polymorphisms related to t
 28 he codons:
 29 238-240 (G80), 520-522 (N174), 742-744 (Q248)
 32 <400> SEQUENCE: 1
 33 atg acc agg gcg gga gat cac aac cgc cag aga gga tgc tgt gga tcc 48
 34 Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser
 35 1 5 10 15
 37 ttg gcc gac tac ctg acc tct gca aaa ttc ctt ctc tac ctt ggt cat 96
 38 Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His
 39 20 25 30
 41 tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg 144
 42 Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val
 43 35 40 45
 45 ttt ctg gta gag ctc tat gga aac agc ctc ctt ttg aca gca gtc tac 192
 46 Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
 47 50 55 60
 49 ggg ctg gtg gtg gca ggg tct gtt ctg gtc ctg gga gcc atc atc ggt 240
 50 Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly
 51 65 70 75 80
 53 gac tgg gtg gac aag aat gct aga ctt aaa gtg gcc cag acc tcg ctg 288
 54 Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu
 55 85 90 95
 57 gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg 336
 58 Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met
 59 100 105 110
 61 gtt ttc tta cat aaa cat gag ctt ctg acc atg tac cat gga tgg gtt 384

62 Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val

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63	115	120	125	
65	ctc act tcc tgc tat atc ctg atc atc act att gca aat att gca aat	432		
66	Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn			
67	130 135 140			
69	ttg gcc agt act gct act gca atc aca atc caa agg gat tgg att gtt	480		
70	Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val			
71	145 150 155 160			
73	gtt gtt gca gga gaa gac aga agc aaa cta gca aat atg aat gcc aca	528		
74	Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr			
75	165 170 175			
77	ata cga agg att gac cag tta acc aac atc tta gcc ccc atg gct gtt	576		
78	Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val			
79	180 185 190			
81	ggc cag att atg aca ttt ggc tcc cca gtc atc ggc tgt ggc ttt att	624		
82	Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile			
83	195 200 205			
85	tcg gga tgg aac ttg gta tcc atg tgc gtg gag tac gtc ctg ctc tgg	672		
86	Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp			
87	210 215 220			
89	aag gtt tac cag aaa acc cca gct cta gct gtg aaa gct ggt ctt aaa	720		
90	Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys			
91	225 230 235 240			
93	gaa gag gaa act gaa ttg aaa cag ctg aat tta cac aaa gat act gag	768		
94	Glu Glu Glu Thr Glu Leu Lys Gln Leu Asn Leu His Lys Asp Thr Glu			
95	245 250 255			
97	cca aaa ccc ctg gag gga act cat cta atg ggt gtg aaa gac tct aac	816		
98	Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn			
99	260 265 270			
101	atc cat gag ctt gaa cat gag caa gag cct act tgt gcc tcc cag atg	864		
102	Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met			
103	275 280 285			
105	gct gag ccc ttc cgt acc ttc cga gat gga tgg gtc tcc tac tac aac	912		
106	Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn			
107	290 295 300			
109	cag cct gtg ttt ctg gct ggc atg ggt ctt gct ttc ctt tat atg act	960		
110	Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr			
111	305 310 315 320			
113	gtc ctg ggc ttt gac tgc atc acc aca ggg tac gcc tac act cag gga	1008		
114	Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly			
115	325 330 335			
117	ctg agt ggt tcc atc ctc agt att ttg atg gga gca tca gct ata act	1056		
118	Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr			
119	340 345 350			
121	gga ata atg gga act gta gct ttt act tgg cta cgt cga aaa tgt ggt	1104		
122	Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly			
123	355 360 365			
125	ttg gtt cgg aca ggt ctg atc tca gga ttg gca cag ctt tcc tgt ttg	1152		
126	Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu			
127	370 375 380			

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129 atc ttg tgt gtg atc tct gta ttc atg cct gga agc ccc ctg gac ttg      1200
130 Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu
131 385      390      395      400
133 tcc gtt tct cct ttt gaa gat atc cga tca agg ttc att caa gga gag      1248
134 Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu
135      405      410      415
137 tca att aca cct acc aag ata cct gaa att aca act gaa ata tac atg      1296
138 Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met
139      420      425      430
141 tct aat ggg tct aat tct gct aat att gtc ccg gag aca agt cct gaa      1344
142 Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu
143      435      440      445
145 tct gtg ccc ata atc tct gtc agt ctg ctg ttt gca ggc gtc att gct      1392
146 Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala
147      450      455      460
149 gct aga atc ggt ctt tgg tcc ttt gat tta act gtg aca cag ttg ctg      1440
150 Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu
151 465      470      475      480
153 caa gaa aat gta att gaa tct gaa aga ggc att ata aat ggt gta cag      1488
154 Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln
155      485      490      495
157 aac tcc atg aac tat ctt ctt gat ctt ctg cat ttc atc atg gtc atc      1536
158 Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile
159      500      505      510
161 ctg gct cca aat cct gaa gct ttt ggc ttg ctc gta ttg att tca gtc      1584
162 Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val
163      515      520      525
165 tcc ttt gtg gca atg ggc cac att atg tat ttc cga ttt gcc caa aat      1632
166 Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn
167      530      535      540
169 act ctg gga aac aag ctc ttt gct tgc ggt cct gat gca aaa gaa gtt      1680
170 Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val
171 545      550      555      560
173 agg aag gaa aat caa gca aat aca tct gtt gtt tga      1716
174 Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val
175      565      570
178 <210> SEQ ID NO: 2
179 <211> LENGTH: 571
180 <212> TYPE: PRT
181 <213> ORGANISM: Homo sapiens
183 <400> SEQUENCE: 2
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186 1      5      10      15
189 Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His
190      20      25      30
193 Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val
194      35      40      45
197 Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
198      50      55      60

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201 Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Gly
202 65 70 75 80
205 Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu
206 85 90 95
209 Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met
210 100 105 110
213 Val Phe Leu His Lys His Glu Leu Leu Thr Met Tyr His Gly Trp Val
214 115 120 125
217 Leu Thr Ser Cys Tyr Ile Leu Ile Ile Thr Ile Ala Asn Ile Ala Asn
218 130 135 140
221 Leu Ala Ser Thr Ala Thr Ala Ile Thr Ile Gln Arg Asp Trp Ile Val
222 145 150 155 160
225 Val Val Ala Gly Glu Asp Arg Ser Lys Leu Ala Asn Met Asn Ala Thr
226 165 170 175
229 Ile Arg Arg Ile Asp Gln Leu Thr Asn Ile Leu Ala Pro Met Ala Val
230 180 185 190
233 Gly Gln Ile Met Thr Phe Gly Ser Pro Val Ile Gly Cys Gly Phe Ile
234 195 200 205
237 Ser Gly Trp Asn Leu Val Ser Met Cys Val Glu Tyr Val Leu Leu Trp
238 210 215 220
241 Lys Val Tyr Gln Lys Thr Pro Ala Leu Ala Val Lys Ala Gly Leu Lys
242 225 230 235 240
245 Glu Glu Glu Thr Glu Leu Lys Gln Leu Asn Leu His Lys Asp Thr Glu
246 245 250 255
249 Pro Lys Pro Leu Glu Gly Thr His Leu Met Gly Val Lys Asp Ser Asn
250 260 265 270
253 Ile His Glu Leu Glu His Glu Gln Glu Pro Thr Cys Ala Ser Gln Met
254 275 280 285
257 Ala Glu Pro Phe Arg Thr Phe Arg Asp Gly Trp Val Ser Tyr Tyr Asn
258 290 295 300
261 Gln Pro Val Phe Leu Ala Gly Met Gly Leu Ala Phe Leu Tyr Met Thr
262 305 310 315 320
265 Val Leu Gly Phe Asp Cys Ile Thr Thr Gly Tyr Ala Tyr Thr Gln Gly
266 325 330 335
269 Leu Ser Gly Ser Ile Leu Ser Ile Leu Met Gly Ala Ser Ala Ile Thr
270 340 345 350
273 Gly Ile Met Gly Thr Val Ala Phe Thr Trp Leu Arg Arg Lys Cys Gly
274 355 360 365
277 Leu Val Arg Thr Gly Leu Ile Ser Gly Leu Ala Gln Leu Ser Cys Leu
278 370 375 380
281 Ile Leu Cys Val Ile Ser Val Phe Met Pro Gly Ser Pro Leu Asp Leu
282 385 390 395 400
285 Ser Val Ser Pro Phe Glu Asp Ile Arg Ser Arg Phe Ile Gln Gly Glu
286 405 410 415
289 Ser Ile Thr Pro Thr Lys Ile Pro Glu Ile Thr Thr Glu Ile Tyr Met
290 420 425 430
293 Ser Asn Gly Ser Asn Ser Ala Asn Ile Val Pro Glu Thr Ser Pro Glu
294 435 440 445
297 Ser Val Pro Ile Ile Ser Val Ser Leu Leu Phe Ala Gly Val Ile Ala

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298      450      455      460
301 Ala Arg Ile Gly Leu Trp Ser Phe Asp Leu Thr Val Thr Gln Leu Leu
302 465      470      475      480
305 Gln Glu Asn Val Ile Glu Ser Glu Arg Gly Ile Ile Asn Gly Val Gln
306      485      490      495
309 Asn Ser Met Asn Tyr Leu Leu Asp Leu Leu His Phe Ile Met Val Ile
310      500      505      510
313 Leu Ala Pro Asn Pro Glu Ala Phe Gly Leu Leu Val Leu Ile Ser Val
314      515      520      525
317 Ser Phe Val Ala Met Gly His Ile Met Tyr Phe Arg Phe Ala Gln Asn
318      530      535      540
321 Thr Leu Gly Asn Lys Leu Phe Ala Cys Gly Pro Asp Ala Lys Glu Val
322 545      550      555      560
325 Arg Lys Glu Asn Gln Ala Asn Thr Ser Val Val
326      565      570

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329 <210> SEQ ID NO: 3

330 <211> LENGTH: 1716

331 <212> TYPE: DNA

332 <213> ORGANISM: Homo sapiens

334 <220> FEATURE:

335 <221> NAME/KEY: CDS

336 <222> LOCATION: (1)..(1716)

337 <223> OTHER INFORMATION: cDNA encoding a ferroportin 1 mutated in position

(G80) .

340 <400> SEQUENCE: 3

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341 atg acc agg gcg gga gat cac aac cgc cag aga gga tgc tgt gga tcc      48
342 Met Thr Arg Ala Gly Asp His Asn Arg Gln Arg Gly Cys Cys Gly Ser
343 1      5      10      15
345 ttg gcc gac tac ctg acc tct gca aaa ttc ctt ctc tac ctt ggt cat      96
346 Leu Ala Asp Tyr Leu Thr Ser Ala Lys Phe Leu Leu Tyr Leu Gly His
347      20      25      30
349 tct ctc tct act tgg gga gat cgg atg tgg cac ttt gcg gtg tct gtg      144
350 Ser Leu Ser Thr Trp Gly Asp Arg Met Trp His Phe Ala Val Ser Val
351      35      40      45
353 ttt ctg gta gag ctc tat gga aac agc ctc ctt ttg aca gca gtc tac      192
354 Phe Leu Val Glu Leu Tyr Gly Asn Ser Leu Leu Leu Thr Ala Val Tyr
355      50      55      60
357 ggg ctg gtg gtg gca ggg tct gtt ctg gtc ctg gga gcc atc atc agt      240
358 Gly Leu Val Val Ala Gly Ser Val Leu Val Leu Gly Ala Ile Ile Ser
359 65      70      75      80
361 gac tgg gtg gac aag aat gct aga ctt aaa gtg gcc cag acc tcg ctg      288
362 Asp Trp Val Asp Lys Asn Ala Arg Leu Lys Val Ala Gln Thr Ser Leu
363      85      90      95
365 gtg gta cag aat gtt tca gtc atc ctg tgt gga atc atc ctg atg atg      336
366 Val Val Gln Asn Val Ser Val Ile Leu Cys Gly Ile Ile Leu Met Met
367      100      105      110
369 gtt ttc tta cat aaa cat gag ctt ctg acc atg tac cat gga tgg gtt      384
370 Val Phe Leu His Lys His Glu Leu Thr Met Tyr His Gly Trp Val
371      115      120      125
373 ctc act tcc tgc tat atc ctg atc atc act att gca aat att gca aat      432

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RAW SEQUENCE LISTING ERROR SUMMARY
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Please Note:

Use of n and/or Xaa have been detected in the Sequence Listing. Please review the Sequence Listing to ensure that a corresponding explanation is presented in the <220>

to <223> fields of each sequence which presents at least one n or Xaa.

Seq#:28; Xaa Pos. 3

Seq#:29; Xaa Pos. 3

Seq#:30; Xaa Pos. 3

VERIFICATION SUMMARY

PATENT APPLICATION: US/10/560,157

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Input Set : A:\Seqlist.txt

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L:9 M:270 C: Current Application Number differs, Replaced Current Application Number

L:1259 M:257 W: Feature value mis-spelled or invalid, <221> Name/Key for SEQ ID#:9

L:1550 M:341 W: (46) "n" or "Xaa" used, for SEQ ID#:28 after pos.:0

L:1567 M:341 W: (46) "n" or "Xaa" used, for SEQ ID#:29 after pos.:0

L:1584 M:341 W: (46) "n" or "Xaa" used, for SEQ ID#:30 after pos.:0